



## otopalatodigital syndrome type 1

Otopalatodigital syndrome type 1 is a disorder primarily involving abnormalities in skeletal development. It is a member of a group of related conditions called otopalatodigital spectrum disorders, which also includes otopalatodigital syndrome type 2, frontometaphyseal dysplasia, and Melnick-Needles syndrome. In general, these disorders involve hearing loss caused by malformations in the tiny bones in the ears (ossicles), problems in the development of the roof of the mouth (palate), and skeletal abnormalities involving the fingers and/or toes (digits).

Otopalatodigital syndrome type 1 is usually the mildest of the otopalatodigital spectrum disorders. People with this condition usually have characteristic facial features including wide-set and downward-slanting eyes; prominent brow ridges; and a small, flat nose. Affected individuals also have hearing loss and chest deformities. They have abnormalities of the fingers and toes, such as blunt, square-shaped (spatulate) fingertips; shortened thumbs and big toes; and unusually long second toes.

Affected individuals may be born with an opening in the roof of the mouth (a cleft palate). They may have mildly bowed limbs, and limited range of motion in some joints. People with otopalatodigital syndrome type 1 may be somewhat shorter than other members of their family. Males with this disorder often have more severe signs and symptoms than do females, who may show only the characteristic facial features.

### Frequency

Otopalatodigital syndrome type 1 is a rare disorder, affecting fewer than 1 in every 100,000 individuals. Its specific incidence is unknown.

### Genetic Changes

Mutations in the *FLNA* gene cause otopalatodigital syndrome type 1.

The *FLNA* gene provides instructions for producing the protein filamin A, which helps build the network of protein filaments (cytoskeleton) that gives structure to cells and allows them to change shape and move. Filamin A binds to another protein called actin, and helps the actin to form the branching network of filaments that make up the cytoskeleton. Filamin A also links actin to many other proteins to perform various functions within the cell.

A small number of mutations in the *FLNA* gene have been identified in people with otopalatodigital syndrome type 1. The mutations all result in changes to the filamin A protein in the region that binds to actin. The mutations responsible for otopalatodigital syndrome type 1 are described as "gain-of-function" because they appear to enhance

the activity of the filamin A protein or give the protein a new, atypical function. Researchers believe that the mutations may change the way the filamin A protein helps regulate processes involved in skeletal development, but it is not known how changes in the protein relate to the specific signs and symptoms of otopalatodigital syndrome type 1.

## **Inheritance Pattern**

This condition is inherited in an X-linked dominant pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In females (who have two X chromosomes), a mutation in one of the two copies of the gene in each cell is sufficient to cause the disorder. In males (who have only one X chromosome), a mutation in the only copy of the gene in each cell causes the disorder. In most cases, males experience more severe symptoms of the disorder than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

## **Other Names for This Condition**

- cranioorodigital syndrome
- faciopalatoosseous syndrome
- FPO
- OPD syndrome, type 1
- oto-palato-digital syndrome, type I
- Taybi syndrome

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Oto-palato-digital syndrome, type I  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265251/>

### Other Diagnosis and Management Resources

- GeneReview: Otopalatodigital Spectrum Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1393>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>

- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Bone Diseases  
<https://medlineplus.gov/bonediseases.html>

### Genetic and Rare Diseases Information Center

- Oto-palato-digital syndrome type 1  
<https://rarediseases.info.nih.gov/diseases/5121/oto-palato-digital-syndrome-type-1>

### Educational Resources

- Disease InfoSearch: Oto-palato-digital syndrome type 1  
<http://www.diseaseinfosearch.org/Oto-palato-digital+syndrome+type+1/5495>
- Orphanet: Otopalatodigital syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=669](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=669)

### Patient Support and Advocacy Resources

- Children's Craniofacial Association  
<http://www.ccakids.com/>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/otopalatodigital-syndrome-type-i-and-ii/>

### GeneReviews

- Otopalatodigital Spectrum Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1393>

### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22otopalatodigital+syndrome+type+1%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28otopalatodigital+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- OTOPALATODIGITAL SYNDROME, TYPE I  
<http://omim.org/entry/311300>

## **Sources for This Summary**

- GeneReview: Otopalatodigital Spectrum Disorders  
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